HB 6580

Testimony for Public Hearing
Public Health Committee
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Chairman Ritter, Chairman Gerratana, Members of the Public Health Committee:

I am writing to request your support for HB6580 An Act to Create an Advisory Council on Rare Diseases. I am the mother of three children who each have a rare disease. My oldest child has MCADD(1) which affects her body's ability to break down fats for energy. My younger two children have mitochondrial disease(2) which is a progressive, multi-system disease with no cure. The incidence of mitochondrial disease is now approaching the frequency of childhood cancer(3). My family has had many struggles with these rare diseases. My oldest daughter almost passed away at three days old but was saved thanks to our state's excellent Newborn Screening program and our pediatrician. The first years raising a child with a rare, life-threatening disorder were extremely difficult. She required round the clock feedings, a feeding tube, monthly blood draws, multiple hospitalizations and many doctor visits. This time was extremely isolating and terrifying as a new mother. When my younger two children were born it was quickly determined that they were not affected by MCADD for which we were so thankful. Unfortunately it soon became clear that they were in fact not "healthy." My son suffered from failure to thrive, hypoglycemia, severe silent reflux, and lactose intolerance requiring extremely expensive prescription formula. He has also been diagnosed with seizures, muscle hypotonia, heat intolerance, anisocoria, encephalopathy, exercise intolerance, Pervasive Development Disorder- Not Otherwise Specified and other diagnosis. When my youngest daughter was born

she had a relatively typical first year but shortly afterwards she also began to lose developmental skills, stopped progressing and also was found to have life-threatening hypoglycemia. She has also suffered from chronic constipation at time requiring hospitalization, hydronephrosis, severe reflux that caused her to be 100% tube fed for a period of time. Local doctors could not determine the cause of my children's various ailments but their symptoms needed to be controlled so both my children underwent surgery for a feeding tube and my son also had skin, muscle and liver biopsies.They both have been hospitalized many times for illness, sometimes concurrently. After many years of being referred to various doctors, most out of state, they were finally diagnosed with mitochondrial disease. The journey to find a diagnosis was fraught with self doubt, fear and a feeling that I was failing my children. There is no pain greater than seeing your child suffer.

Despite having medically complex children my number one priority has always been to give them as normal a life as possible. These diseases do not and will not define them. Due to the wonderful care my daughter received in her early years she is now thriving and is a bright, happy, social nine year old that participates in ballet, gymnastics, ice skating, summer camps, girl scouts and much more. However I always have the thought in the back of my mind that she does have a life-long, life-threatening disorder that will always need to be managed. The future is less certain for my younger children and they are much more limited in the activities they can do and my son struggles every day in school. Despite this I try to keep my family's focus on living life to it's fullest. However the hardships I face on a daily basis are sometimes overwhelming. Paying the copays and deductibles for the medical care my children require can be a huge burden. Several of the medicines prescribed for my children are not covered by insurance because they are considered over-the-counter and I have not been able to afford them. When my son transitioned from

preschool to elementary school we had to fight and hire an advocate to get the services he required. Due to the tube feedings my children require finding a caregiver is difficult and only their father and myself are able to watch them overnight. Finding medical care for my youngest children has been nothing short of a nightmare. We recently lost their specialist when Tufts Floating Hospital in Boston closed their metabolic clinic down. This is one of 5 major mitochondrial disease clinics to close in the last year which has affected thousands of patients(4). Currently my children do not have a specialist and we will have to travel quite far at great expense to find a new one. While we have a very competent local team to care for them, none of them have expertise in mitochondrial disease.

My family is not alone in this struggle. There are over 6,800 rare diseases affecting an estimated 25 to 30 million americans.(5) There is currently no registry in Connecticut to track the incidence of these rare diseases. The creation of an Advisory Council on Rare Diseases would benefit my family and all affected families, adults and their providers. The Council would determine the incidence of rare disease in Connecticut and allow various experts, providers and affected people to come together to determine the common issues faced and identify available resources and streamline them or reduce redundancies, assist in raise awareness and identify private funding sources. Individually our voices are small but combine we can be heard and make a difference.

Thank you for your time and consideration.

Heather Harwood

Sources:

1- http://ghr.nlm.nih.gov/condition/medium-chain-acyl-coa-dehydrogenase-deficiency

2-http://www.umdf.org/site/c.8qKOJ0MvF7LUG/b.7934627/k.3711/What_is_Mitochondrial_Disease.htm

3- http://www.umdf.org/site/pp.aspx?c=8qKOJ0MvF7LUG&b=7934639

4 -http://www.mitoaction.org/files/MitoAction-ClinicClosings-Statement.pdf

5- http://www.genome.gov/27531963